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Chronic granulomatous disease (CGD)

What is CGD?

In CGD one type of immune cell, called neutrophils, do not work properly. This leads to increased susceptibility to infections, and in particular infections caused by bacteria and fungi. CGD is present from birth and the symptoms usually start within the first 2 years of life.

CGD is a genetic disease and can be inherited in two ways. The most common type is X-linked. This means that it usually only affects boys, though females can be carriers of the condition. Less commonly, CGD is inherited in an autosomal recessive manner. This means that both males and females can be affected, and both males and females can be carriers.

Why does it happen?

Neutrophils are an important first line of defence for the immune system and they attack bacterial and fungal infections. They do this by consuming the organisms causing infection and then killing them. In CGD, a genetic mutation means that the neutrophils are unable to kill bacteria and fungi properly, and therefore the ability to defend against these infections is reduced.

What might my symptoms be?

The symptoms in CGD result from the infections. Abscesses form which affect the skin, but can also affect internal organs such as the liver. Other infections can affect the chest or gut. The exact symptoms will depend on the infection(s) present at the time but may include sweats, fever, tiredness, cough, sputum production, muscle pains and/or diarrhoea. The severity of CGD is variable, but without appropriate treatment the lifespan of affected individuals is reduced.

How is CGD diagnosed?

In an individual with symptoms suggestive of CGD a blood test of neutrophil function is performed. This checks the ability of the neutrophils to kill bacteria and fungi. If abnormal then this suggests CGD. Further genetic tests on blood may be performed to determine the precise type of CGD. This allows information about the inheritance of this condition to be given (ie genetic counselling).

What treatments can be used?

Infections need prompt treatment with antibiotics. Antibiotics may also be given long term to try to prevent any new infections. Despite this, the infections often prove difficult to treat. Surgical drainage of abscesses may be required. Sometimes a product called interferon is given as in some patients this can boost the killing ability of the neutrophils.
However, all of the above treatments, while useful to help fight off infections, do not treat the underlying cause. For this reason a 'curative' treatment such as bone marrow transplant may be considered. Gene therapy for the one of the types of CGD may be an option in selected cases.

Further information

The above information is a summary of what effects CGD might have, but different patients may have different symptoms. Please ask us in clinic any questions that you have about CGD and/or your treatment.
PID UK (Primary Immunodeficiency UK) and UKPIPS (UK Primary Immunodeficiency Patient Support) are patient groups for those affected by immunodeficiencies. Their websites are below (note that we have no editorial control for the websites and take no responsibility for their content):
www.piduk.org
www.ukpips.org.uk

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Last revised: September 2018
Next review: September 2021